



## ABHD5 gene

abhydrolase domain containing 5

### Normal Function

The *ABHD5* gene provides instructions for making a protein that turns on (activates) an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides, which are the main source of stored energy in cells. Triglycerides are the major component of cell structures called lipid droplets (also called adiposomes). The ABHD5 protein and the ATGL enzyme are found on the surface of lipid droplets. Once activated, the ATGL enzyme breaks down triglycerides in these structures to provide energy for the body.

### Health Conditions Related to Genetic Changes

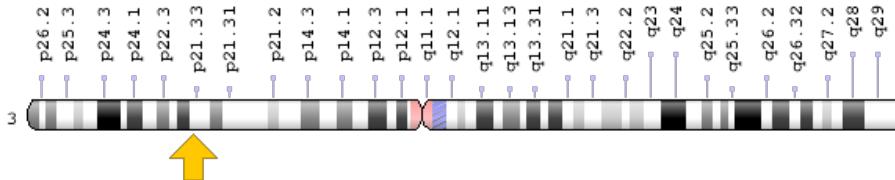
#### Chanarin-Dorfman syndrome

At least 20 mutations in the *ABHD5* gene have been found to cause Chanarin-Dorfman syndrome. These mutations impair the ABHD5 protein's ability to activate the ATGL enzyme. Without an active ATGL enzyme, triglycerides within lipid droplets cannot be broken down. As a result, these fats accumulate in various organs and tissues throughout the body, causing the signs and symptoms of Chanarin-Dorfman syndrome.

### Chromosomal Location

Cytogenetic Location: 3p21.33, which is the short (p) arm of chromosome 3 at position 21.33

Molecular Location: base pairs 43,690,883 to 43,722,725 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## **Other Names for This Gene**

- ABHD5\_HUMAN
- CDS
- CGI-58
- CGI58
- CGI58 protein
- IECN2
- MGC8731
- NCIE2

## **Additional Information & Resources**

### Genetic Testing Registry

- GTR: Genetic tests for ABHD5  
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51099%5Bgeneid%5D>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ABHD5%5BTIAB%5D%29+OR+%28%28CGI58%5BTIAB%5D%29+OR+%28CGI-58%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

### OMIM

- ABHYDROLASE DOMAIN-CONTAINING 5  
<http://omim.org/entry/604780>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ABHD5.html](http://atlasgeneticsoncology.org/Genes/GC_ABHD5.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ABHD5%5Bgene%5D>
- HGNC Gene Family: Abhydrolase domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/4>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=21396](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21396)

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51099>
- UniProt  
<http://www.uniprot.org/uniprot/Q8WTS1>

## Sources for This Summary

- OMIM: ABHYDROLASE DOMAIN-CONTAINING 5  
<http://omim.org/entry/604780>
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